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Case Report

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Aicardi Syndrome: A Rare Childhood Disorder

Luthfor Rahman*[1], AKM Mamunur Rashid[1]

1 Department of Pediatrics, Naogaon Medical College, Naogaon

***Corresponding author**:
Dr. Luthfor Rahman
Associate Professor, Department of Pediatrics, Naogaon Medical College

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ABSTRACT: Aicardi syndrome represents a complex disorder that affects many systems and is typically associated with agenesis of the corpus callosum, distinctive chorioretinal lacunae, and infantile spasms. Patients are almost all female, suggesting a genetic abnormality of the X chromosome (it may be lethal in males during fetal life). The presenting case was an 8 months 21 days old girl who had complaints of convulsion which started at the age of 3 months. The convulsion was generalized and tonic in nature and occurred 3 to 4 times daily with an up rolling of eyeball and lasted for 5 minutes. Post-ictally the child becomes active. It was not associated with fever or vomiting. On examination there were markedly delayed developmental milestones, tour shaped forehead, small head, closed fontanelles and cranial sutures, depressed nasal bridge, frequent protrusion of tongue and high arched palate. On investigation, CT scan of the brain showed partial agenesis of corpus callosum and bilateral ventriculomegaly. EEG reports are consistent with severe epileptic encephalopathy with modified hypsarrhythmia, and karyotyping report was normal. Drug choice was vigabatrin which is not available in our country. The patient was treated with Inj. ACTH for 3 months and oral levetiracetam and phenobarbitone. Seizure was controlled only for 3months during ACTH injection thereafter, frequent seizure episode.

Keywords: Aicardi Syndrome, Agenesis of Corpus Callosum, Hypsarrhythmia.

INTRODUCTION

Aicardi syndrome is defined by the triad of infantile spasms, agenesis of the corpus callosum and chorioretinal lacunae.[1] Patients are almost all female suggesting a genetic abnormality of X chromosome (it may be lethal in males during fetal life). Seizures become evident during the first few months and are typically resistant to anticonvulsants.[2] We present a case of Aicardi syndrome in an 8 months 21 days old girl, along with a brief review of this rare syndrome to create awareness.

CASE REPORT

Suraiya, an 8 months 21 days old incompletely immunized girl of non-consanguineous parents of poor socioeconomic family, born at term by NVD at Ad-din medical college hospital, Khulna, hailing from Doulotpur, Khulna got admitted at pediatric ward, Khulna Medical college Hospital (KMCH) on 13th February 2020 with the complaints of convulsion since the age of 3months.It was generalized and tonic in nature and occurred 3 to 4 times daily with up rolling eye ball and lasted for 5 minutes. Post-ictally the child becomes active. It was not associated with fever or vomiting. Suraiya was not growing well in comparison with her peer; grandmother added. On query the grandmother told Suraiya was fed expressed breast milk and infant formula due to refusal of suckling since birth. There was no history of contact with TB patients, loose motion, evening pyrexia or night sweats. With these complaints Suraiya got admitted at KMCH two times at the age of 4th and 6th months and was treated as bronchopneumonia with microcephaly and was referred

to National Institute of Neurosciences (NINS). Thereafter, the child was shifted to DMCH and Infectious Disease Hospital (IDH), Mohakhali Dhaka where she was prescribed with antibiotic along with Inj. ACTH and levetiracetam oral. There was no history of such type of illness at the family. The mother was under regular antenatal cheek up and during post—natal period the baby was admitted at Khulna Shishu Hospital with the complaints of excessive crying. There was evidence of markedly delayed developmental milestones. On examination the baby was mildly pale, non-cteric, not cyanosed, non-edematous. BCG scar mark present. There is a tour shaped forehead, depressed nasal bridge, frequent protrusion of tongue, high arched palate, closed fontanelles and cranial sutures. Her vital signs are within normal limits. Anthropometry showed head circumference 33 cm (<-3SD), weight 8kg, length 51 cm and WLZ (-1 to 0 SD). Nervous system examination revel, atrophy of limbs muscles, with increased tone and exaggerated deep tendon reflex and positive Babinski's sign. Higher psychic function could not be evaluated. The examination of cranial nerves found normal with an intact sensation. All other systems reveal no abnormalities.

Investigation

CT scan of the brain showed partial agenesis of corpus callosum and bilateral ventriculomegaly. EEG reports are consistent with severe epileptic encephalopathy with modified hyps arrhythmia and karyotyping report was normal. So, the child was diagnosed as Aiicardi syndrome with global developmental delay with microcephaly with infantile spasms. The drug of choice of infantile spasms is Vigabatrin, which is not available in our country and has retinal toxicity. Treatment with Inj. ACTH 150 units/m^2 divided into twice daily intramuscular injections administered over a 2-wks period with a subsequent gradual tapering over a 2-wk period. Oral levetiracetam 20mg/kg in two divided doses and phenobarbitone 4mg/kg/day bid doses was also given.

DISCUSSSION

Aicardi syndrome is a rare genetic disease that was described for the first time in 1965, by Aicardi *et al.*, who studied 117 cases of spasm in infants and discovered eight new cases that comprised this syndrome.[3] Aicardi syndrome (AS) is a rare neurodevelopmental disorder characterized by the classic triad of agenesis of the corpus callosum, chorioretinal lacunae, and infantile spasms. Additional major features consist of cortical malformations, periventricular/subcortical heterotopia, intracranial cysts, and optic nerve coloboma or hypoplasia. Examination findings include microcephaly, axial hypotonia, and appendicular hypertonia along with spasticity. Seizures tend to develop before the age of three months and may become refractory thereafter. Aicardi syndrome is an X-linked dominant condition with lethality in males – therefore affecting primarily girls; the exact genetic basis of Aicardi syndrome is yet to be identified.

The most frequent neurological alterations, which start in the first year of life, include tonic convulsive or limited clonic spasms. The spasms are predominantly asymmetrical, with hemiparesis or hemiplegia on the side that is more affected. These spasms are usually the first manifestation for screening of Aicardi syndrome. The cranial circumference at birth may be within the limits of normality, although a certain degree of microcephaly may be observed.[7] Classical craniofacial features in Aicardi syndrome that have been described include a small philtrum with upturned nasal tip and decreased angle of the nasal bridge, big ears, prominent premaxilla and sparse eyebrows on lateral aspects, but these were not present in our case.[8] Skeletal anomalies such as fused vertebrae, hemi-vertebrae, blocked vertebrae and absent ribs could be present, which may lead to scoliosis.[9] Imaging diagnostic methods may reveal conditions ranging from classical findings like partial or total agenesis of the corpus callosum to other findings likeventricular dilatation, hydrocephaly and cortical heterotopy.[10]

Magnetic resonance imaging (MRI) is the gold-standard examination for diagnosing this syndrome. MRI is the imaging method used for confirming the diagnosis, although transfontanellar ultrasonography can also be chosen for screening, confirming the diagnosis and follow-up, with similar results. Ultrasound is relatively inexpensive and accessible; there are no contraindications, and the images are obtained non-invasively.[11] Aicardi syndrome carries a bad prognosis due to its association with medically intractable seizures and mental retardation.[12] Management includes anti-seizure treatment, physical, occupational, speech, vision therapy, and orthopedic assessment and treatment.

CONCLUSSION

As Aicardi syndrome carries a bad prognosis due to its association with medically intractable seizures and mental retardation early diagnosis and multi-disciplinary approach needed for better management.

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